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DEPARTMENT OF COMMERCE

National Institute of Standards and Technology

Genome in a Bottle Consortium – Progress and Planning Workshop

AGENCY: National Institute of Standards & Technology (NIST), Commerce.

ACTION: Notice of public workshop.

SUMMARY: NIST announces the Genome in a Bottle (GIAB) Consortium meeting to be held on Thursday and Friday, September 15 and 16, 2016. The Genome in a Bottle Consortium is developing the reference materials, reference methods, and reference data needed to assess confidence in human whole genome variant calls. A principal motivation for this consortium is to enable performance assessment of sequencing and science-based regulatory oversight of clinical sequencing. The purpose of this meeting is to update participants about progress of the consortium work, continue to get broad input from individual stakeholders to update or refine the consortium work plan, continue to broadly solicit consortium membership from interested stakeholders, and invite members to participate in work plan implementation. September 15 will be a new sample thinkshop to discuss new GIAB genomes in parallel with a data jamboree to develop high-confidence calls

for difficult variants and difficult regions. September 16 will be the plenary session to present GIAB progress updates and emerging technical work.

DATES: The Genome in a Bottle Consortium meeting will be held on Thursday, September 15, 2016 from 9:00 AM to 5:30 PM Eastern Time and Friday, September 16, 2016 from 8:30 AM to 2:00 PM Eastern Time. Attendees must register by 5:00 PM Eastern Time on Thursday, September 8, 2016.

ADDRESSES: The meeting will be held in Lecture Room A, Lecture Room B, and the Green Auditorium, Building 101, National Institute of Standards and Technology, 100 Bureau Drive, Gaithersburg, MD 20899. Please note admittance instructions under the SUPPLEMENTARY INFORMATION section of this notice.

FOR FURTHER INFORMATION CONTACT: For further information contact Justin Zook by email at jzook@nist.gov or by phone at (301) 975-4133 or Marc Salit by email at salit@nist.gov or by phone at (650) 350-2338. To register, go to: https://appam.certain.com/profile/form/index.cfm?PKformID=0x311041593.

SUPPLEMENTARY INFORMATION: Clinical application of ultra-high throughput sequencing for hereditary genetic diseases and oncology is rapidly growing. At present, there are no widely accepted genomic standards or quantitative performance metrics for confidence in variant calling. These standards and quantitative performance metrics are needed to achieve the confidence in measurement results expected for sound, reproducible research and regulated applications in the clinic. On April 13, 2012, NIST convened the

workshop “Genome in a Bottle” to initiate a consortium to develop the reference materials, reference methods, and reference data needed to assess confidence in human whole genome variant calls (www.genomeinabottle.org). On August 16-17, 2012, NIST hosted the first large public meeting of the Genome in a Bottle Consortium, with about 100 participants from government, academic institutions, and industry. This meeting was announced in the Federal Register (77 FR 43237) on July 24, 2012. A principal motivation for this consortium is to enable science-based regulatory oversight of clinical sequencing.

At the August 2012 meeting, the consortium established work plans for four technical working groups with the following responsibilities:

(1) Reference Material (RM) Selection and Design: select appropriate sources for whole genome RMs and identify or design synthetic DNA constructs that could be spiked-in to samples for measurement assurance.

(2) Measurements for Reference Material Characterization: design and carry out experiments to characterize the RMs using multiple sequencing methods, other methods, and validation of selected variants using orthogonal technologies.

(3) Bioinformatics, Data Integration, and Data Representation: develop methods to analyze and integrate the data for each RM, as well as select appropriate formats to represent the data.

(4) Performance Metrics and Figures of Merit: develop useful performance metrics and figures of merit that can be obtained through measurement of the RMs.

The products of these technical working groups will be a set of well-characterized whole genome and synthetic DNA RMs along with the methods (documentary standards) and reference data necessary for use of the RMs. These products will be designed to help enable translation of whole genome sequencing to regulated clinical applications. The pilot NIST whole genome RM 8398 was released in May 2015 and is available at http://tinyurl.com/giabpilot. The consortium is currently analyzing and integrating data from two trios that are candidate NIST RMs. The consortium meets in workshops two times per year, in January at Stanford University in Palo Alto, CA, and in August at the National Institute of Standards and Technology in Gaithersburg, MD. At these workshops, including the last meetings at Stanford in January 2016 and at NIST in August 2015, participants in the consortium have discussed progress in developing well-characterized genomes for NIST Reference Materials and planned future experiments and analysis of these genomes (see https://federalregister.gov/a/2012-18064, https://federalregister.gov/a/2013-18934, https://federalregister.gov/a/2014-18841, https://federalregister.gov/a/2015-01158, and https://www.federalregister.gov/articles/2016/01/05/2015-33140/genome-in-a-bottle-consortium-progress-and-planning-workshop for announcements of past workshops at NIST and Stanford). The January 2016 meeting was announced in the Federal Register (81 FR 226) on January 5, 2016, and the meeting is summarized at https://docs.google.com/document/d/1VdP96SYCPcZZvXprowMq8rp6FURCxSh1uo4Dd1tTpJY/edit?usp=drive_web.

There is no cost for participating in the consortium. No proprietary information will be shared as part of the consortium, and all research results will be in the public domain.

All attendees are required to pre-register. Anyone wishing to attend this meeting must pre-register at https://appam.certain.com/profile/form/index.cfm?PKformID=0x311041593 by 5:00 PM Eastern Time on Thursday, September 8, 2016, in order to attend.

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